



What is it?

Points To Remember About Osteopetrosis

- In osteopetrosis, bones become too dense but can easily break. This happens when new bone grows faster than old bone is removed.
- Bone that is too dense can cause serious problems throughout the body.
- Treatments for osteopetrosis depend on the type and how severe the disease is.
- Your doctor will advise you to practice healthy habits that include a balanced diet and proper dental care.

In osteopetrosis, bones are too dense but easily broken. This happens when new bone grows faster than old bone is removed.

Bone that is too dense can cause serious problems throughout the body. It can also cause problems with bone marrow (tissue inside the bones). This can lead to low levels of cells needed to fight infection, carry oxygen to the body's cells, or control bleeding.

Who gets it?

You need to have a gene from one or both parents to have the disorder.

What are the types?

Types of osteopetrosis include:

- A severe form that is seen at or shortly after birth.
- A less severe form that is found in children younger than age 10.
- The mildest form, which may not be diagnosed until adolescence or adulthood.

What are the symptoms?

The most common symptoms of osteopetrosis are:

- Bone fractures.
- Low blood cell levels.
- Vision loss.
- Hearing loss.
- Dental problems related to infection.

What causes it?

Osteopetrosis is caused by problems in one or more genes involved in breaking down bone tissue. This causes new bone to grow faster than old bone can be removed, making the bone too thick.

In some cases, one or both of your parents passed the problem gene to you. In many cases, you didn't get the gene from your parents. In this case, the problem gene may have appeared when you were conceived.

Is there a test for it?

Your doctor may do the following to test you for osteopetrosis and problems related to the disease:

- Take pictures of your bones.
- Hearing and vision tests.
- Blood tests.

How is it treated?

Treatments for osteopetrosis depend on the type and how severe the disease is and can include:

- Medications, nutritional supplements, or hormones to stop the disease from getting worse

or help with its symptoms.

- Physical and occupational therapy to help children develop motor and other skills.
- Treatment for fractures.
- Regular doctor visits to monitor the eye, ear, nose, and throat.
- Bone marrow transplant: In children with the severe form of the disease. This can stop the disease from getting worse, although it can't fix damage that already occurred.

Who treats it?

Several types of health care professionals may treat you, including:

- Hematologists, who are blood disease specialists.
- Endocrinologists, who specialize in hormonal and metabolic disorders.
- Orthopaedists, who treat and perform surgery for bone and joint diseases.
- Ophthalmologists, who specialize in eye care.
- Otolaryngologists, who are ear, nose, and throat (ENT) specialists.

Living with it

In addition to supervising your medical treatments, your doctor will advise you to practice healthy habits in your everyday life. These include:

- Eating a balanced diet to support normal growth and development.
- Practicing good dental care to decrease the chance of dental infections.

For More Info

U.S. Food and Drug Administration

Toll free: 888-INFO-FDA (888-463-6332)

Website: <https://www.fda.gov>

Drugs@FDA at <https://www.accessdata.fda.gov/scripts/cder/daf/>. Drugs@FDA is a searchable catalog of FDA-approved drug products.

Centers for Disease Control and Prevention, National Center for Health Statistics

Website: <https://www.cdc.gov/nchs>

NIH Osteoporosis and Related Bone Diseases ~ National Resource Center

Website: <https://www.bones.nih.gov>

Genetic and Rare Diseases Information Center

Website: <https://rarediseases.info.nih.gov/GARD/>

Genetics Home Reference

Website: <https://www.ghr.nlm.nih.gov/condition/osteopetrosis>

American Academy of Orthopaedic Surgeons

Website: <https://www.aaos.org> (con información en español)

Orphanet

Website: <https://www.orpha.net>

St. Jude Children's Research Hospital

Website: <https://www.stjude.org>

If you need more information about available resources in your language or other languages, please visit our webpages below or contact the NIAMS Information Clearinghouse at NIAMSInfo@mail.nih.gov.

- [Asian Language Health Information](#)
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